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Tripatara et al. Archives of biochemistry and biophysics (1999 Jul 1) 367(1)39-50

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Bonne et al. Pediatric research (1993 Mar) 33(3)284-8

Dahl et al. Human Genetics (1991 May) 87(1) 49-53

Huh et al. Journal of Biological Chemistry (1990 Aug 5) 265 (22) 13320-6

Kitano et al. Journal of inhereted metabolic disease (1989) 12(2)91-107.

Juliet Einsmann
AU 1634
703 306 5824
Office: CM1-12D15
Mailbox: CM1-12E12

Teach that $E_{1\alpha}$ subunit mutations are related to severity of $E_{1\alpha}$ deficiency in patients with PDH complex deficiency. Are silent, however as to the molecular basis of the mutation.

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Teach a series of mutations that cause changes in coding sequence of PDH E1a gene (table 2) in patients w/ PDH complex deficiency

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Absence of a complex in

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